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L1 near10 fabry adj disease	0

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L2	L1 near10 fabry adj disease	0	L2
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FILE 'MEDLINE, CAPLUS, BIOSIS, SCISEARCH' ENTERED AT 17:50:30 ON 10 FEB 2004

L1 5686 S LYSOSOM?(3A)HYDROLASE
L2 4040 S LYSOSOMAL(W)HYDROLASE
L3 1 S L2(10A)FABRY(W)DISEASE

=> d bib ab l3

L3 ANSWER 1 OF 1 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
AN 1988:374756 BIOSIS
DN PREV198886058666; BA86:58666
TI STRUCTURAL ORGANIZATION OF THE HUMAN ALPHA GALACTOSIDASE A GENE FURTHER EVIDENCE FOR THE ABSENCE OF A 3' UNTRANSLATED REGION.
AU BISHOP D F [Reprint author]; KORNREICH R; DESNICK R J
CS DIV MED GENETICS, MOUNT SINAI SCH MED, 100TH ST AND FIFTH AVE, NEW YORK, NY 10029, USA
SO Proceedings of the National Academy of Sciences of the United States of America, (1988) Vol. 85, No. 11, pp. 3903-3907.
CODEN: PNASA6. ISSN: 0027-8424.
DT Article
FS BA
LA ENGLISH
ED Entered STN: 18 Aug 1988
Last Updated on STN: 18 Aug 1988
AB Human α-galactosidase A (α-D-galactoside galactohydrolase; EC 3.2.1.22) is a lysosomal hydrolase encoded by a gene localized to the chromosomal region Xq22. The deficient activity of this enzyme results in Fabry disease, an X chromosome-linked recessive disorder that leads to premature death in affected males. For studies of the structure and function of α-galactosidase A and for characterization of the genetic lesions in families with Fabry disease, the full-length cDNA was isolated, sequenced, and used to screen human genomic libraries. The 1393-base-pair full-length cDNA had a 60-nucleotide 5' untranslated region and encoded a precursor peptide of 429 amino acids including a signal peptide of 31 residues. Three overlapping λ clones spanning 32 kilobases were identified that contained the entire ≈12-kilobase chromosomal gene as well as ≈9 and ≈11 kilobases of 5' and 3' flanking sequence, respectively. The gene had seven exons. The genomic exonic and full-length cDNA sequences were identical. All intron-exon splice junctions conformed to the GT/AT consensus sequence. The 5' flanking region of this lysosomal housekeeping gene contained Sp1 and CCAAT box promoter elements as well as sequences corresponding to the activator protein 1 (AP1), octanucleotide ("OCTA"), and "core" enhancer elements. There was an upstream "HTF" island (Hpa II tiny fragments) followed by four direct repeats of the "chorion box" enhancer. The unique lack of a 3' untranslated sequence in the α-galactosidase A cDNA was confirmed by sequencing additional cDNA clones and the genomic 3' region.

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